- Search History will be lost after eight hours of inactivity.
- Search numbers may not be continuous; all searches are represented.
- To save search indefinitely, click query # and select Save in My NCBI.
- To combine searches use #search, e.g., #2 AND #3 or click query # for more options.

Search	Most Recent Queries	Time	Result
#9	Select **7587391**	18:35:29	1
#4	**Related Articles** for **PubMed** (Select **2517620**)	18:35:03	110
#8	Select **3** document(s)	18:33:42	3
#7	Select **2517620**	18:32:06	1
#6	Select **4** document(s)	18:11:43	4
#5	Select **2** document(s)	18:09:42	2

Clear History

Dec 18 2006 06:34:27

☐ **1:** P31327. Reports ...[gi:4033707]

Comment Features Sequence

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LOCUS       P31327                  1500 aa            linear   PRI 12-DEC-2006
DEFINITION  Carbamoyl-phosphate synthase [ammonia], mitochondrial precursor
            (Carbamoyl-phosphate synthetase I) (CPSase I).
ACCESSION   P31327
VERSION     P31327  GI:4033707
DBSOURCE    swissprot: locus CPSM_HUMAN, accession P31327;
            class: standard.
            extra accessions:O43774,Q7Z5I5
            created: Jul 1, 1993.
            sequence updated: Dec 15, 1998.
            annotation updated: Dec 12, 2006.
            xrefs: D90282.1, BAA14328.1, Y15793.1, CAA75785.1, AF154830.1,
            AAD38072.1, AY317138.1, AAP84318.1, AY167007.1, AAO31763.1,
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            AY167005.1, AY167006.1, AF536523.1, AAN77181.1, BX640601.1,
            CAE45707.1, JQ1348
            xrefs (non-sequence databases): UniGene:Hs.149252, HSSP:P00968,
            IntAct:P31327, GermOnline:ENSG00000021826, Ensembl:ENSG00000021826,
            KEGG:hsa:1373, HGNC:2323, HPA:CAB003781, MIM: 237300, MIM: 608307,
            BioCyc:MetaCyc:MONOMER-11364, Reactome:REACT_13.1,
            ArrayExpress:P31327, RZPD-ProtExp:IOH12892, GO:0005739, GO:0004087,
            GO:0005515, InterPro:IPR011761, InterPro:IPR013816,
            InterPro:IPR006275, InterPro:IPR006274, InterPro:IPR001317,
            InterPro:IPR002474, InterPro:IPR005483, InterPro:IPR005480,
            InterPro:IPR005481, InterPro:IPR005479, InterPro:IPR011702,
            InterPro:IPR000991, InterPro:IPR011607, InterPro:IPR013817,
            Pfam:PF00289, Pfam:PF02786, Pfam:PF02787, Pfam:PF00988,
            Pfam:PF00117, Pfam:PF02142, PRINTS:PR00098, PRINTS:PR00099,
            PRINTS:PR00096, TIGRFAMs:TIGR01369, TIGRFAMs:TIGR01368,
            PROSITE:PS50975, PROSITE:PS00866, PROSITE:PS00867
KEYWORDS    Acetylation; Allosteric enzyme; Alternative splicing; ATP-binding;
            Disease mutation; Ligase; Mitochondrion; Nucleotide-binding;
            Polymorphism; Repeat; Transit peptide; Urea cycle.
SOURCE      Homo sapiens (human)
  ORGANISM  Homo sapiens
            Eukaryota; Metazoa; Chordata; Craniata; Vertebrata; Euteleostomi;
            Mammalia; Eutheria; Euarchontoglires; Primates; Haplorrhini;
            Catarrhini; Hominidae; Homo.
REFERENCE   1  (residues 1 to 1500)
  AUTHORS   Haraguchi,Y., Uchino,T., Takiguchi,M., Endo,F., Mori,M. and
            Matsuda,I.
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TITLE Cloning and sequence of a cDNA encoding human carbamyl phosphate synthetase I: molecular analysis of hyperammonemia
JOURNAL Gene 107 (2), 335-340 (1991)
PUBMED 1840546
REMARK NUCLEOTIDE SEQUENCE [MRNA] (ISOFORM 1), AND VARIANTS SER-1266; LEU-1283 AND ASN-1406.
TISSUE=Liver

REFERENCE 2 (residues 1 to 1500)
AUTHORS Finckh,U., Kohlschutter,A., Schafer,H., Sperhake,K., Colombo,J.P. and Gal,A.
TITLE Prenatal diagnosis of carbamoyl phosphate synthetase I deficiency by identification of a missense mutation in CPS1
JOURNAL Hum. Mutat. 12 (3), 206-211 (1998)
PUBMED 9711878
REMARK NUCLEOTIDE SEQUENCE [MRNA] (ISOFORM 1), VARIANT CPS1 DEFICIENCY MET-544, AND VARIANT ALA-344.
TISSUE=Liver

REFERENCE 3 (residues 1 to 1500)
AUTHORS Summar,M.L., Hall,L.D., Eeds,A.M., Hutcheson,H.B., Kuo,A.N., Willis,A.S., Rubio,V., Arvin,M.K., Schofield,J.P. and Dawson,E.P.
TITLE Characterization of genomic structure and polymorphisms in the human carbamyl phosphate synthetase I gene
JOURNAL Gene 311, 51-57 (2003)
PUBMED 12853138
REMARK NUCLEOTIDE SEQUENCE [MRNA] (ISOFORM 1), AND VARIANTS ALA-344; SER-1376 AND ASN-1406.

REFERENCE 4 (residues 1 to 1500)
AUTHORS Huo,R., Zhu,H., Huang,X.Y., Xu,Z.Y., Lu,L., Xu,M., Yin,L.L., Li,J.M., Zhou,Z.M. and Sha,J.H.
TITLE Direct Submission
JOURNAL Submitted (??-JUN-2003)
REMARK NUCLEOTIDE SEQUENCE (ISOFORM 2).
TISSUE=Testis

REFERENCE 5 (residues 1 to 1500)
AUTHORS Funghini,S., Donati,M.A., Pasquini,E., Zammarchi,E. and Morrone,A.
TITLE Structural organization of the human carbamyl phosphate synthetase I gene (CPS1) and identification of two novel genetic lesions
JOURNAL Hum. Mutat. 22 (4), 340-341 (2003)
PUBMED 12955727
REMARK NUCLEOTIDE SEQUENCE (ISOFORM 1), VARIANTS CPS1 DEFICIENCY GLY-457 AND ARG-810, AND VARIANT ASN-1406.

REFERENCE 6 (residues 1 to 1500)
AUTHORS Haeberle,J., Schmidt,E., Pauli,S., Rapp,B., Christensen,E., Wermuth,B. and Koch,H.G.
TITLE Gene structure of human carbamylphosphate synthetase 1 and novel mutations in patients with neonatal onset
JOURNAL Hum. Mutat. 21, 444-444 (2003)
PUBMED 12655559
REMARK NUCLEOTIDE SEQUENCE [GENOMIC DNA] (ISOFORM 1), AND VARIANTS CPS1 DEFICIENCY SER-843 AND GLU-875.

REFERENCE 7 (residues 1 to 1500)
CONSRTM The German cDNA consortium
TITLE Direct Submission
JOURNAL Submitted (??-AUG-2003)
REMARK NUCLEOTIDE SEQUENCE [LARGE SCALE MRNA] OF 795-1500.
TISSUE=Small intestine

REFERENCE 8 (residues 1 to 1500)
AUTHORS Aoshima,T., Kajita,M., Sekido,Y., Kikuchi,S., Yasuda,I., Saheki,T., Watanabe,K., Shimokata,K. and Niwa,T.
TITLE Novel mutations (H337R and 238-362del) in the CPS1 gene cause carbamoyl phosphate synthetase I deficiency
JOURNAL Hum. Hered. 52 (2), 99-101 (2001)
PUBMED 11474210
REMARK VARIANT CPS1 DEFICIENCY ARG-337.

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REFERENCE   9  (residues 1 to 1500)
  AUTHORS   Pearson,D.L., Dawling,S., Walsh,W.F., Haines,J.L., Christman,B.W.,
            Bazyk,A., Scott,N. and Summar,M.L.
  TITLE     Neonatal pulmonary hypertension--urea-cycle intermediates, nitric
            oxide production, and carbamoyl-phosphate synthetase function
  JOURNAL   N. Engl. J. Med. 344 (24), 1832-1838 (2001)
   PUBMED   11407344
  REMARK    VARIANT ASN-1406.
COMMENT     On or before Mar 15, 2005 this sequence version replaced gi:87018,
            gi:399296.
            [FUNCTION] Involved in the urea cycle of ureotelic animals where
            the enzyme plays an important role in removing excess ammonia from
            the cell.
            [CATALYTIC ACTIVITY] 2 ATP + NH(3) + CO(2) + H(2)O = 2 ADP +
            phosphate + carbamoyl phosphate.
            [ENZYME REGULATION] Requires N-acetylglutamate as an allosteric
            activator.
            [INTERACTION] P10398:ARAF; NbExp=3; IntAct=EBI-536811, EBI-365961;
            P04049:RAF1; NbExp=2; IntAct=EBI-536811, EBI-365996.
            [SUBCELLULAR LOCATION] Mitochondrion.
            [ALTERNATIVE PRODUCTS] Event=Alternative splicing; Named
            isoforms=2; Name=1; IsoId=P31327-1; Sequence=Displayed; Name=2;
            IsoId=P31327-2; Sequence=VSP_009332; Note=No experimental
            confirmation available.
            [TISSUE SPECIFICITY] Primarily in the liver and small intestine.
            [DOMAIN] The type-1 glutamine amidotransferase domain is defective.
            [DISEASE] Defects in CPS1 are the cause of CPS1 deficiency
            [MIM:237300]; an autosomal recessive metabolic disorder that cause
            a type of hyperammonemia. Clinical symptoms are vomiting in
            infancy, protein intolerance, intermittent ataxia, seizures,
            lethargy, and mental retardation.
            [SIMILARITY] Contains 2 ATP-grasp domains.
            [SIMILARITY] Contains 1 type-1 glutamine amidotransferase domain.
            [WEB RESOURCE] NAME=GeneReviews;
            URL='http://www.genetests.org/query?gene=CPS1'.
FEATURES             Location/Qualifiers
     source          1..1500
                     /organism="Homo sapiens"
                     /db_xref="taxon:9606"
     gene            1..1500
                     /gene="CPS1"
     Protein         1..1500
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                     /note="Mitochondrion (By similarity)."
     Region          39..1500
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                     /experiment="experimental evidence, no additional details
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                /note="Anthranilate phosphoribosyltransferase homolog."
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                /gene="CPS1"
                /region_name="CarA"
                /note="Carbamoylphosphate synthase small subunit [Amino
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                metabolism]; COG0505"
                /db_xref="CDD:30851"
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                /region_name="CPSase_sm_chain"
                /note="Carbamoyl-phosphate synthase small chain, CPSase
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                /db_xref="CDD:41060"
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                /inference="non-experimental evidence, no additional
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                /note="N6-acetyllysine (By similarity)."
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                /experiment="experimental evidence, no additional details
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                /note="A -> S (in Ref. 1)."
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                /site_type="modified"
                /inference="non-experimental evidence, no additional
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     Region     220..410
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                /region_name="Region of interest in the sequence"
                /experiment="experimental evidence, no additional details
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                /note="Glutamine amidotransferase-like."
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                /region_name="GATase1_CPSase"
                /note="This group of sequences represents the small chain
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                synthase, CPSase II; cd01744"
                /db_xref="CDD:28856"
     Region     279
                /gene="CPS1"
                /region_name="Conflict"
                /experiment="experimental evidence, no additional details
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                /note="R -> Q (in Ref. 1)."
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                /db_xref="CDD:40385"
     Region     437..822
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                /region_name="CarB"
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                /experiment="experimental evidence, no additional details
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                /site_type="modified"
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                /note="N6-acetyllysine (By similarity)."
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                /region_name="Variant"
                /experiment="experimental evidence, no additional details
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     Region     718..722
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                /region_name="Conflict"
                /experiment="experimental evidence, no additional details
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                /note="RLSRS -> KMSPN (in Ref. 1)."
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                /region_name="Conflict"
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/note="A -> T (in Ref. 1)."
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/note="E -> G (in Ref. 1)."
Region 810
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/region_name="Variant"
/experiment="experimental evidence, no additional details recorded"
/note="Q -> R (in CPS1 deficiency). /FTId=VAR_017563."
Region 839..962
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/note="L -> S (in CPS1 deficiency). /FTId=VAR_017564."
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/region_name="Variant"
/experiment="experimental evidence, no additional details recorded"
/note="K -> E (in CPS1 deficiency). /FTId=VAR_017565."
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                /note="GD -> EN (in Ref. 1)."
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                /note="M -> L (in dbSNP:rs1047887). /FTId=VAR_017567."
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                /experiment="experimental evidence, no additional details
                recorded"
                /note="A -> V (in Ref. 1)."
     Region     1360..1475
                /gene="CPS1"
                /region_name="MGS_CPS_I_III"
                /note="Methylglyoxal synthase-like domain found in pyr1
                and URA1-like carbamoyl phosphate synthetases (CPS),
                including ammonia-dependent CPS Type I, and
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                /db_xref="CDD:29635"
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      361 negimheskp ffavqfhpev tpgpidteyl fdsffslikk gkattitsvl pkpalvasrv
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      781 fhgtssrigs smksvgevma igrtfeesfq kalrmchpsi egftprlpmn kewpsnldlr
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     1261 srsfpfvskt lgvdfidvat kvmigenvde khlptldhpi ipadyvaika pmfswprlrd
     1321 adpilrcema stgevacfge gihtaflkam lstgfkipqk giligiqqsf rprflgvaeq
     1381 lhnegfklfa teatsdwlna nnvpatpvaw psqegqnpsl ssirklirdg sidlvinlpn
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//
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